

Opis choroby *

Definicja

X-linked creatine transporter deficiency (CRTR-D) is a creatine deficiency syndrome characterized clinically by global developmental delay/ intellectual disability (DD/ID) with prominent speech/language delay, autistic behavior and seizures.

Dane

Klasyfikacja	Synonimy
Choroba	Creatine transporter deficiency Niedobór transportera kreatyny SLC6A8 deficiency SLC6A8 deficiency

Kod ORPHA 52503	Kod OMIM 300352	Kod ICD10 E72.8
Kod ICD11 5C53.4		

*Źródło

orphanet